

Hypermethioninemia (HMET)

An amino acid disorder

What is it?

Hypermethioninemia (also known as HMET) is an inherited amino acid disorder. People with amino acid disorders, like HMET, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical (enzyme). Since the body cannot properly break down the protein, certain amino acids build up in the blood and urine and cause problems when a person eats normal amounts of protein.

What are the symptoms?

People with HMET usually have no symptoms. There have been reports of people with HMET having foul breath, and having problems with the insulation surrounding the brain. People with HMET typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

HMET is inherited in an autosomal recessive manner. This means that for a person to be affected with HMET, he or she must have inherited two non-working copies of the gene responsible for causing HMET. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have HMET. Typically, there is no family history of HMET in an affected person. The number of people affected with HMET is unknown..

How is it detected?

HMET can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

HMET can be treated by eating a diet low in protein and they may be given a special formula, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

Save Babies Through Screening Foundation

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(610) 993-0545 Email: email@savebabies.org

Website: <http://www.savebabies.org/diseasedescriptions.php>